



## COQ4 gene

coenzyme Q4

### Normal Function

The COQ4 gene provides instructions for making a protein that is involved in the production of a molecule called coenzyme Q10, although its specific role in this process is unknown. Research suggests that the COQ4 protein may help organize other proteins involved in coenzyme Q10 production into a stable functional group (a protein complex).

Coenzyme Q10 has several critical functions in cells throughout the body. In cell structures called mitochondria, coenzyme Q10 plays an essential role in a process called oxidative phosphorylation, which converts the energy from food into a form cells can use. Coenzyme Q10 is also involved in producing pyrimidines, which are building blocks of DNA, its chemical cousin RNA, and molecules such as ATP and GTP that serve as energy sources in the cell. In cell membranes, coenzyme Q10 acts as an antioxidant, protecting cells from damage caused by unstable oxygen-containing molecules (free radicals), which are byproducts of energy production.

### Health Conditions Related to Genetic Changes

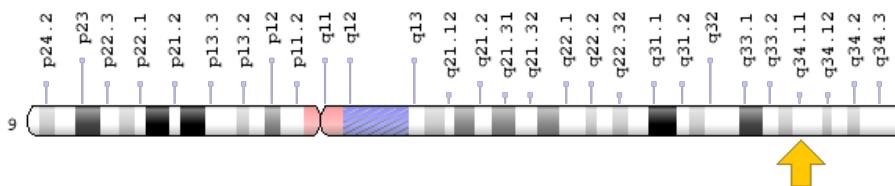
#### primary coenzyme Q10 deficiency

At least 12 mutations in the COQ4 gene have been found to cause a disorder known as primary coenzyme Q10 deficiency. This rare disease usually becomes apparent in infancy or early childhood, but it can occur at any age. It can affect many parts of the body, most often the brain, muscles, and kidneys. The COQ4 gene mutations associated with this disorder greatly reduce or eliminate the production of the COQ4 protein, which prevents the normal production of coenzyme Q10. Studies suggest that a shortage (deficiency) of coenzyme Q10 impairs oxidative phosphorylation and increases the vulnerability of cells to damage from free radicals. A deficiency of coenzyme Q10 may also disrupt the production of pyrimidines. These changes can cause cells throughout the body to malfunction, which may help explain the variety of organs and tissues that can be affected by primary coenzyme Q10 deficiency.

## Chromosomal Location

Cytogenetic Location: 9q34.11, which is the long (q) arm of chromosome 9 at position 34.11

Molecular Location: base pairs 128,322,486 to 128,334,072 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CGI-92
- coenzyme Q biosynthesis protein 4 homolog
- coenzyme Q4 homolog
- COQ10D7
- ubiquinone biosynthesis protein COQ4 homolog, mitochondrial isoform 1 precursor
- ubiquinone biosynthesis protein COQ4 homolog, mitochondrial isoform 2 precursor

## Additional Information & Resources

### Educational Resources

- Linus Pauling Institute: Coenzyme Q10  
<http://lpi.oregonstate.edu/mic/dietary-factors/coenzyme-Q10>
- Molecular Biology of the Cell (fourth edition, 2002): How Cells Obtain Energy From Food  
<https://www.ncbi.nlm.nih.gov/books/NBK26882/>
- The Cell: A Molecular Approach (second edition, 2000): The Mechanism of Oxidative Phosphorylation  
<https://www.ncbi.nlm.nih.gov/books/NBK9885/>

### GeneReviews

- Primary Coenzyme Q10 Deficiency  
<https://www.ncbi.nlm.nih.gov/books/NBK410087>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28COQ4%5BTIAB%5D%29+OR+%28coenzyme+Q4%5BTIAB%5D%29%29+OR+%28%28coenzyme+biosynthesis+protein+4+homolog%5BTIAB%5D%29+OR+%28coenzyme+Q4+homolog%5BTIAB%5D%29+OR+%28ubiquinone+biosynthesis+protein+COQ4+homolog,+mitochondrial+isoform+1+precursor%5BTIAB%5D%29+OR+%28ubiquinone+biosynthesis+protein+COQ4+homolog,+mitochondrial+isoform+2+precursor%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

## OMIM

- COENZYME Q4, S. CEREVISIAE, HOMOLOG OF  
<http://omim.org/entry/612898>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=COQ4%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=19693](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19693)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/51117>
- UniProt  
<http://www.uniprot.org/uniprot/Q9Y3A0>

## **Sources for This Summary**

- Acosta MJ, Vazquez Fonseca L, Desbats MA, Cerqua C, Zordan R, Trevisson E, Salviati L. Coenzyme Q biosynthesis in health and disease. *Biochim Biophys Acta*. 2016 Aug;1857(8):1079-85. doi: 10.1016/j.bbabiobio.2016.03.036. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/27060254>
- Brea-Calvo G, Haack TB, Karall D, Ohtake A, Invernizzi F, Carrozzo R, Kremer L, Dusi S, Fauth C, Scholl-Bürgi S, Graf E, Ahting U, Resta N, Laforgia N, Verrigni D, Okazaki Y, Kohda M, Martinelli D, Freisinger P, Strom TM, Meitinger T, Lamperti C, Lacson A, Navas P, Mayr JA, Bertini E, Murayama K, Zeviani M, Prokisch H, Ghezzi D. COQ4 mutations cause a broad spectrum of mitochondrial disorders associated with CoQ10 deficiency. *Am J Hum Genet*. 2015 Feb 5;96(2):309-17. doi: 10.1016/j.ajhg.2014.12.023.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25658047>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4320255/>
- OMIM: COENZYME Q4, S. CEREVISIAE, HOMOLOG OF  
<http://omim.org/entry/612898>

- Casarin A, Jimenez-Ortega JC, Trevisson E, Pertegato V, Doimo M, Ferrero-Gomez ML, Abbadi S, Artuch R, Quinzii C, Hirano M, Basso G, Ocaña CS, Navas P, Salviati L. Functional characterization of human COQ4, a gene required for Coenzyme Q10 biosynthesis. *Biochem Biophys Res Commun.* 2008 Jul 18;372(1):35-9. doi: 10.1016/j.bbrc.2008.04.172.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18474229>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4345104/>
- Chung WK, Martin K, Jalas C, Braddock SR, Juusola J, Monaghan KG, Warner B, Franks S, Yudkoff M, Lulis L, Rhodes RH, Prasad V, Torti E, Cho MT, Shinawi M. Mutations in COQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. *J Med Genet.* 2015 Sep;52(9):627-35. doi: 10.1136/jmedgenet-2015-103140.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26185144>
- Desbats MA, Lunardi G, Doimo M, Trevisson E, Salviati L. Genetic bases and clinical manifestations of coenzyme Q10 (CoQ 10) deficiency. *J Inher Metab Dis.* 2015 Jan;38(1):145-56. doi: 10.1007/s10545-014-9749-9. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25091424>
- Doimo M, Desbats MA, Cerqua C, Cassina M, Trevisson E, Salviati L. Genetics of coenzyme q10 deficiency. *Mol Syndromol.* 2014 Jul;5(3-4):156-62. doi: 10.1159/000362826.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25126048>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4112527/>
- GeneReview: Primary Coenzyme Q10 Deficiency  
<https://www.ncbi.nlm.nih.gov/books/NBK410087>
- Salviati L, Trevisson E, Rodriguez Hernandez MA, Casarin A, Pertegato V, Doimo M, Cassina M, Agosto C, Desbats MA, Sartori G, Sacconi S, Memo L, Zuffardi O, Artuch R, Quinzii C, Dimauro S, Hirano M, Santos-Ocaña C, Navas P. Haploinsufficiency of COQ4 causes coenzyme Q10 deficiency. *J Med Genet.* 2012 Mar;49(3):187-91. doi: 10.1136/jmedgenet-2011-100394.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22368301>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3983946/>

---

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/COQ4>

Reviewed: April 2017

Published: April 25, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services